



ABCA12 gene

ATP binding cassette subfamily A member 12

Normal Function

The *ABCA12* gene provides instructions for making a protein known as an ATP-binding cassette (ABC) transporter. ABC transporter proteins carry many types of molecules across cell membranes. In particular, the *ABCA12* protein plays a major role in transporting fats (lipids) in cells that make up the outermost layer of skin (the epidermis). This lipid transport appears to be essential for normal development of the skin. The *ABCA12* protein is also found in several other tissues, including the testes, placenta, lung, stomach, and fetal brain and liver.

Health Conditions Related to Genetic Changes

harlequin ichthyosis

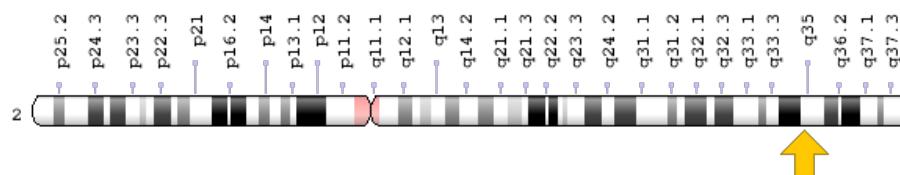
Several mutations in the *ABCA12* gene have been identified in people with harlequin ichthyosis. Most of these mutations probably lead to an absence of *ABCA12* protein or the production of an extremely small version of the protein that cannot transport lipids properly. A lack of lipid transport causes numerous problems with the development of the epidermis before and after birth. Specifically, it prevents the skin from forming an effective barrier against fluid loss (dehydration) and infections, and leads to the formation of hard, thick scales characteristic of harlequin ichthyosis.

lamellar ichthyosis

Chromosomal Location

Cytogenetic Location: 2q35, which is the long (q) arm of chromosome 2 at position 35

Molecular Location: base pairs 214,931,542 to 215,138,591 on chromosome 2 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- ABCAC_HUMAN
- ATP-binding cassette 12
- ATP-binding cassette transporter 12
- ATP-binding cassette, sub-family A (ABC1), member 12
- ATP-binding cassette, sub-family A, member 12
- ICR2B

Additional Information & Resources

Educational Resources

- The Human ATP-Binding Cassette (ABC) Transporter Superfamily: ABCA Genes
<https://www.ncbi.nlm.nih.gov/books/NBK3/#A166>

GeneReviews

- Autosomal Recessive Congenital Ichthyosis
<https://www.ncbi.nlm.nih.gov/books/NBK1420>

Genetic Testing Registry

- GTR: Genetic tests for ABCA12
<https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=26154%5Bgeneid%5D>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28ABCA12%5BALL%5D%29+OR+%28ICR2B%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

OMIM

- ATP-BINDING CASSETTE, SUBFAMILY A, MEMBER 12
<http://omim.org/entry/607800>
- ICHTHYOSIS, CONGENITAL, AUTOSOMAL RECESSIVE 4A
<http://omim.org/entry/601277>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_ABCA12.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=ABCA12%5Bgene%5D>

- HGNC Gene Family: ATP binding cassette subfamily A
<http://www.genenames.org/cgi-bin/genefamilies/set/805>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=14637
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/26154>
- UniProt
<http://www.uniprot.org/uniprot/Q86UK0>

Sources for This Summary

- Akiyama M, Sakai K, Sugiyama-Nakagiri Y, Yamanaka Y, McMillan JR, Sawamura D, Niizeki H, Miyagawa S, Shimizu H. Compound heterozygous mutations including a de novo missense mutation in ABCA12 led to a case of harlequin ichthyosis with moderate clinical severity. *J Invest Dermatol.* 2006 Jul;126(7):1518-23. Epub 2006 May 4.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/16675967>
- Akiyama M, Sugiyama-Nakagiri Y, Sakai K, McMillan JR, Goto M, Arita K, Tsuji-Abe Y, Tabata N, Matsuoka K, Sasaki R, Sawamura D, Shimizu H. Mutations in lipid transporter ABCA12 in harlequin ichthyosis and functional recovery by corrective gene transfer. *J Clin Invest.* 2005 Jul;115(7):1777-84.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/16007253>
Free article on PubMed Central: <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1159149/>
- Akiyama M. Pathomechanisms of harlequin ichthyosis and ABCA transporters in human diseases. *Arch Dermatol.* 2006 Jul;142(7):914-8. Review.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/16847209>
- Annilo T, Shulnenin S, Chen ZQ, Arnould I, Prades C, Lemoine C, Maintoux-Larois C, Devaud C, Dean M, Denèfle P, Rosier M. Identification and characterization of a novel ABCA subfamily member, ABCA12, located in the lamellar ichthyosis region on 2q34. *Cytogenet Genome Res.* 2002;98(2-3):169-76.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/12697999>
- Hovnanian A. Harlequin ichthyosis unmasked: a defect of lipid transport. *J Clin Invest.* 2005 Jul;115(7):1708-10. Review.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/16007249>
Free article on PubMed Central: <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1159155/>
- Kelsell DP, Norgett EE, Unsworth H, Teh MT, Cullup T, Mein CA, Dopping-Hepenstal PJ, Dale BA, Tadini G, Fleckman P, Stephens KG, Sybert VP, Mallory SB, North BV, Witt DR, Sprecher E, Taylor AE, Ilchyshyn A, Kennedy CT, Goodeye H, Moss C, Paige D, Harper JI, Young BD, Leigh IM, Eady RA, O'Toole EA. Mutations in ABCA12 underlie the severe congenital skin disease harlequin ichthyosis. *Am J Hum Genet.* 2005 May;76(5):794-803. Epub 2005 Mar 8.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/15756637>
Free article on PubMed Central: <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1199369/>

- Lefévre C, Audebert S, Jobard F, Bouadjar B, Lakhdar H, Boughdene-Stambouli O, Blanchet-Bardon C, Heilig R, Foglio M, Weissenbach J, Lathrop M, Prud'homme JF, Fischer J. Mutations in the transporter ABCA12 are associated with lamellar ichthyosis type 2. *Hum Mol Genet*. 2003 Sep 15;12(18):2369-78. Epub 2003 Jul 15.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/12915478>
- Thomas AC, Cullup T, Norgett EE, Hill T, Barton S, Dale BA, Sprecher E, Sheridan E, Taylor AE, Wilroy RS, DeLozier C, Burrows N, Goodyear H, Fleckman P, Stephens KG, Mehta L, Watson RM, Graham R, Wolf R, Slavotinek A, Martin M, Bourn D, Mein CA, O'Toole EA, Kelsell DP. ABCA12 is the major harlequin ichthyosis gene. *J Invest Dermatol*. 2006 Nov;126(11):2408-13. Epub 2006 Aug 10.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/16902423>
- Thomas AC, Sinclair C, Mahmud N, Cullup T, Mellerio JE, Harper J, Dale BA, Turc-Carel C, Hohl D, McGrath JA, Vahlquist A, Hellstrom-Pigg M, Ganemo A, Metcalfe K, Mein CA, O'Toole EA, Kelsell DP. Novel and recurring ABCA12 mutations associated with harlequin ichthyosis: implications for prenatal diagnosis. *Br J Dermatol*. 2008 Mar;158(3):611-3. Epub 2007 Nov 6.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/17986308>
- Uitto J. The gene family of ABC transporters--novel mutations, new phenotypes. *Trends Mol Med*. 2005 Aug;11(8):341-3. Review.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/15996518>

Reprinted from Genetics Home Reference:

<https://ghr.nlm.nih.gov/gene/ABCA12>

Reviewed: November 2008

Published: February 14, 2017

Lister Hill National Center for Biomedical Communications
U.S. National Library of Medicine
National Institutes of Health
Department of Health & Human Services